SMN2 gene

survival of motor neuron 2, centromeric

Normal Function

The *SMN2* gene provides instructions for making the survival motor neuron (SMN) protein. The SMN protein is found throughout the body, with high levels in the spinal cord. This protein is particularly important for the maintenance of specialized nerve cells called motor neurons, which are located in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem). Motor neurons control muscle movements.

Several different versions of the SMN protein are produced from the *SMN2* gene, but only one version (called isoform d) is full size and fully functional. The other versions are smaller and easily broken down. The full-size protein made from the *SMN2* gene is identical to the protein made from a similar gene called *SMN1*; however, much less full-size SMN protein is produced from the *SMN2* gene compared with the *SMN1* gene.

In cells, the SMN protein plays an important role in processing molecules called messenger RNA (mRNA), which serve as genetic blueprints for making proteins. Messenger RNA begins as a rough draft (pre-mRNA) and goes through several processing steps to become a final, mature form. The SMN protein helps to assemble the cellular machinery needed to process pre-mRNA. The SMN protein may have additional functions in nerve cells. Research findings indicate that the SMN protein is important for the development of specialized outgrowths from nerve cells called dendrites and axons. Dendrites and axons are required for the transmission of impulses from nerve to nerve and from nerves to muscles.

Health Conditions Related to Genetic Changes

spinal muscular atrophy

Typically, people have two copies of the *SMN1* gene and up to two copies of the *SMN2* gene in each cell. In people with spinal muscular atrophy, both copies of the *SMN1* gene are altered or missing. In some cases, individuals have three or more copies of the *SMN2* gene. In those with spinal muscular atrophy, additional copies of the *SMN2* gene are associated with a milder course of the disorder.

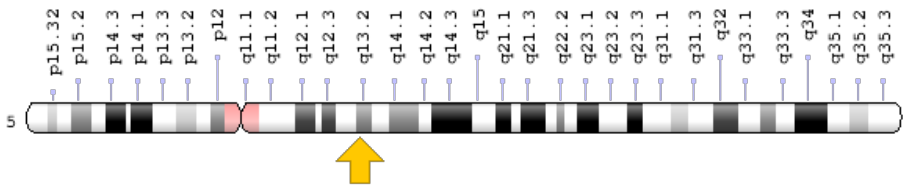
When both copies of the *SMN1* gene are altered or missing, little or no SMN protein is produced from this gene. Extra *SMN2* genes can help replace some of the SMN protein that is lost due to mutations in the *SMN1* genes. The symptoms of spinal muscular atrophy still occur, however, because only a small amount of the full-size SMN protein is produced from the *SMN2* genes. In general, symptoms are less

severe and begin later in life in affected individuals with three or more copies of the *SMN2* gene compared with those who have two copies of this gene.

Chromosomal Location

Cytogenetic Location: 5q13.2, which is the long (q) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 70,049,523 to 70,077,595 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BCD541
- C-BCD541
- centromeric SMN
- SMN_HUMAN
- SMNC

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Collection: Proteins that Help with the Formation of RNA-Protein Complexes
  https://www.ncbi.nlm.nih.gov/books/NBK6016/#A43536
- Molecular Biology of the Cell (4th edition, 2002): The Nucleus Contains a Variety of Subnuclear Structures
  https://www.ncbi.nlm.nih.gov/books/NBK26887/#A1048

GeneReviews

- Spinal Muscular Atrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1352

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SMN2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- SURVIVAL OF MOTOR NEURON 2
  http://omim.org/entry/601627

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SMN2%5Bgene%5D
- HGNC Gene Family: Tudor domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/780
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11118
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6607
- UniProt
  http://www.uniprot.org/uniprot/Q16637

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